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(54) Title: METHODS AND COMPOSITIONS FOR ASSAYING MUTATIONS IN NUCLEIC ACIDS AND THEIR USES IN DIAGNOSIS OF GENETIC DISEASES AND CANCERS

(57) Abstract: The present invention relates to a method for assaying the presence or the absence of at least one mutation on a strand of nucleic acid paired in a duplex form comprising at least the steps of contacting said duplex with at least one compound able to undergo a specific base pairing interaction with suspected mismatch and assaying for said mismatch by an analytical method. The invention further relates to the use in the diagnosis of predisposition to genetic diseases and cancers and in the diagnosis and prognosis of said diseases and cancers, like human breast cancer. The invention also relates to compositions including a compound able to undergo specific base pairing interaction, in association with a DNA fragment having a nucleic sequence relating to a gene on which point mutation(s) has been associated or putatively associated with a genetic disease or an increased predisposition to said disease.